ARID1A gene

AT-rich interaction domain 1A

Normal Function

The *ARID1A* gene provides instructions for making a protein that forms one piece (subunit) of several different SWI/SNF protein complexes. SWI/SNF complexes regulate gene activity (expression) by a process known as chromatin remodeling. Chromatin is the network of DNA and protein that packages DNA into chromosomes. The structure of chromatin can be changed (remodeled) to alter how tightly DNA is packaged. Chromatin remodeling is one way gene expression is regulated during development; when DNA is tightly packed, gene expression is lower than when DNA is loosely packed.

Through their ability to regulate gene activity, SWI/SNF complexes are involved in many processes, including repairing damaged DNA; copying (replicating) DNA; and controlling the growth, division, and maturation (differentiation) of cells. The ARID1A protein and other SWI/SNF subunits are thought to act as tumor suppressors, which keep cells from growing and dividing too rapidly or in an uncontrolled way.

The ARID1A subunit is able to attach (bind) to DNA and is thought to help target SWI/SNF complexes to the chromatin location that needs to be remodeled.

Health Conditions Related to Genetic Changes

cholangiocarcinoma

Coffin-Siris syndrome

At least three mutations in the *ARID1A* gene can cause Coffin-Siris syndrome. This condition is characterized by delayed development, abnormalities of the fifth (pinky) fingers or toes, and characteristic facial features that are described as coarse. The *ARID1A* gene mutations involved in Coffin-Siris syndrome lead to an abnormally short, nonfunctional protein. As a result, affected individuals have half the normal amount of functioning ARID1A protein. Although it is unclear how these changes affect SWI/SNF complexes, researchers suggest that *ARID1A* gene mutations result in abnormal chromatin remodeling. Disturbance of this process alters the activity of many genes and disrupts several cellular processes, which could explain the diverse signs and symptoms of Coffin-Siris syndrome. People with Coffin-Siris syndrome do not appear to have an increased risk of cancer (see below).

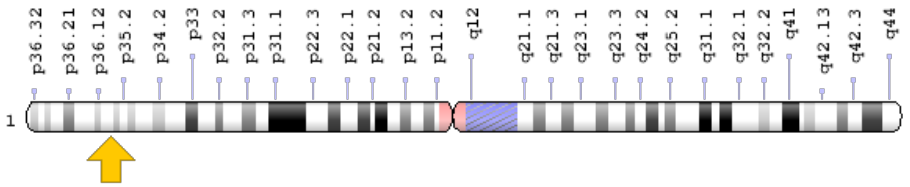
cancers

Mutations in the *ARID1A* gene have been found in many types of cancer, including cancers of the ovaries and lining of the uterus (endometrium) in women and cancers of the kidney, stomach, bladder, lung, breast, and brain. These mutations are somatic, which means they are acquired during a person's lifetime and are present only in tumor cells. The mechanism by which mutations in the *ARID1A* gene contribute to cancer is unknown, although it is thought that changes in SWI/SNF complexes are involved. These changes may impair normal cell differentiation, which leads to the overgrowth of certain cell types, causing cancer. Alternatively, abnormal SWI/SNF complexes may disrupt the regulation of genes that help control the growth and division of cells, which leads to cancer. It is likely that other genetic changes in addition to *ARID1A* gene mutations are necessary for cancer development.

Chromosomal Location

Cytogenetic Location: 1p36.11, which is the short (p) arm of chromosome 1 at position 36.11

Molecular Location: base pairs 26,696,031 to 26,782,110 on chromosome 1 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ARI1A_HUMAN
- ARID domain-containing protein 1A
- AT rich interactive domain 1A (SWI-like)
- AT-rich interactive domain-containing protein 1A
- B120
- BAF250
- BAF250a
- BM029

- brain protein 120
- BRG1-associated factor 250a
- C1orf4
- chromatin remodeling factor p250
- ELD
- hELD
- hOSA1
- MRD14
- osa homolog 1
- OSA1
- OSA1 nuclear protein
- P270
- SMARCF1
- SWI-like protein
- SWI/SNF complex protein p270
- SWI/SNF-related, matrix-associated, actin-dependent regulator of chromatin subfamily F member 1

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): ATP-Driven Chromatin Remodeling Machines Change Nucleosome Structure
  https://www.ncbi.nlm.nih.gov/books/NBK26834/#A644
- Molecular Biology of the Cell (fourth edition, 2002): Chromosomal DNA and Its Packaging in the Chromatin Fiber
  https://www.ncbi.nlm.nih.gov/books/NBK26834/

GeneReviews

- Coffin-Siris Syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK131811

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28ARID1A%5BTI%5D%29+OR+%28AT+rich+interactive+domain+1A%5BTI%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D

OMIM

- AT-RICH INTERACTION DOMAIN-CONTAINING PROTEIN 1A
  http://omim.org/entry/603024

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/ARID1AID44231ch1p36.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=ARID1A%5Bgene%5D
- HGNC Gene Family: AT-rich interaction domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/418
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11110
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/8289
- UniProt
  http://www.uniprot.org/uniprot/O14497

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